CS460Project

June 26, 2024

1 CS460

1.1 Destiny Tudara

1.1.1 Adapting the Burrows-Wheeler Transform (BWT) for Personalized DNA Analysis

[ ]:
```
import pandas as pd
import os
import matplotlib.pyplot as plt

# Step 1: Read the 23andMe raw data
def read_23andme(file_path):
    with open(file_path, 'r') as file:
        lines = file.readlines()
        data_lines = [line.strip() for line in lines if not line.
 ↪startswith('#')]
    data = [line.split('\t') for line in data_lines]
    df = pd.DataFrame(data, columns=['rsid', 'chromosome', 'position',␣
 ↪'genotype'])
    return df

# Step 2: Function to concatenate rsid and genotype to form the encoded string
def encode_genetic_data(df):
    genetic_string = ''.join(df['rsid'] + df['genotype'])
    return genetic_string

# Step 3: Function to perform BWT
def bwt_transform(s):
    s = s + "$"  # Add end of string marker
    n = len(s)
    table = sorted(s[i:] + s[:i] for i in range(n))  # Table of rotations
    return "".join(row[-1] for row in table)  # Last column of the table

# Step 4: Function to perform inverse BWT
def inverse_bwt(r):
    n = len(r)
    table = [""] * n  # Initialize table of n empty strings
```

```
    for _ in range(n):
        table = sorted(r[i] + table[i] for i in range(n))  # Prepend r to each string and sort
    s = [row for row in table if row.endswith("$")][0]  # Find the correct row
    return s.rstrip("$")  # Remove end of string marker

# Main process
file_path = 'rawdata.txt'  # Ensure this file is in the current directory

# Check if the file exists
if os.path.exists(file_path):
    print("File found. Reading the file...")
    df = read_23andme(file_path)
    print("First 5 rows of the genetic data:\n", df.head())

    # Step 2: Encode the genetic data
    genetic_string = encode_genetic_data(df)
    print("Encoded Genetic String (first 100 chars):", genetic_string[:100])

    # Step 3: Apply BWT to the genetic string
    bwt_result = bwt_transform(genetic_string)
    print("BWT Result (first 100 chars):", bwt_result[:100])  # Print first 100 characters of the BWT result for brevity

    # Step 4: Perform inverse BWT to validate
    inverse_result = inverse_bwt(bwt_result)
    print("Inverse BWT Result (first 100 chars):", inverse_result[:100])  # Print first 100 characters to check correctness

else:
    print(f"File not found: {file_path}")
```

```
File found. Reading the file…
First 5 rows of the genetic data:
          rsid chromosome position genotype
0  rs548049170          1    69869       TT
1    rs9283150          1   565508       AA
2  rs116587930          1   727841       GG
3    rs3131972          1   752721       GG
4   rs12184325          1   754105       CC
Encoded Genetic String (first 100 chars): rs548049170TTrs9283150AArs116587930GGr
s3131972GGrs12184325CCrs12567639AArs114525117GGrs12124819AGrs1
```

[1]:
```
import pandas as pd
import os

def read_23andme(file_path):
```

```
    with open(file_path, 'r') as file:
        lines = file.readlines()
        data_lines = [line.strip() for line in lines if not line.
 ↪startswith('#')]
    data = [line.split('\t') for line in data_lines]
    df = pd.DataFrame(data, columns=['rsid', 'chromosome', 'position',␣
 ↪'genotype'])
    return df

# Example usage
file_path = 'rawdata.txt'  # Ensure this file is in the current directory

# Check if the file exists
if os.path.exists(file_path):
    df = read_23andme(file_path)
    print(df.head())
else:
    print(f"File not found: {file_path}")

# List of SNPs associated with AMD
amd_snps = {
    'rs1061170': 'C',  # CFH
    'rs3753394': 'A',  # CFH
    'rs10490924': 'T',  # ARMS2
    'rs2230199': 'G',  # C3
    'rs9332739': 'C',  # C2
    'rs641153': 'A'  # CFB
}

# Filter the DataFrame to include only relevant SNPs
amd_data = df[df['rsid'].isin(amd_snps.keys())].copy()

# Determine if the risk allele is present
amd_data['risk_allele'] = amd_data.apply(lambda row: amd_snps[row['rsid']] in␣
 ↪row['genotype'], axis=1)
amd_data['risk_score'] = amd_data['risk_allele'].astype(int)  # Convert True/
 ↪False to 1/0

# Calculate the overall risk score
overall_risk_score = amd_data['risk_score'].sum()
print(f'Overall AMD Risk Score: {overall_risk_score}')

amd_data
```

```
          rsid chromosome position genotype
0  rs548049170          1    69869       TT
1    rs9283150          1   565508       AA
```

```
2  rs116587930          1   727841       GG
3    rs3131972          1   752721       GG
4   rs12184325          1   754105       CC
Overall AMD Risk Score: 4
```

[1]:

	rsid	chromosome	position	genotype	risk_allele	risk_score
36170	rs3753394	1	196620917	CC	False	0
36181	rs1061170	1	196659237	CC	True	1
231919	rs9332739	6	31903804	CG	True	1
380763	rs10490924	10	124214448	TT	True	1
563627	rs2230199	19	6718387	GG	True	1

1.2 Breast Cancer Gene Analysis

This analysis aims to evaluate the genetic risk for breast cancer based on significant SNPs associated with the BRCA1 and BRCA2 genes. The SNPs included in this analysis are:

1. **BRCA1 (Breast Cancer 1)**
   - **rs799917**
   - **rs1799950**
   - **rs16941**
2. **BRCA2 (Breast Cancer 2)**
   - **rs11571833**
   - **rs144848**

The alleles for these SNPs will be used to predict the likelihood of increased breast cancer risk.

[18]:

```python
import pandas as pd
import os

def read_23andme(file_path):
    with open(file_path, 'r') as file:
        lines = file.readlines()
        data_lines = [line.strip() for line in lines if not line.
 ↪startswith('#')]
    data = [line.split('\t') for line in data_lines]
    df = pd.DataFrame(data, columns=['rsid', 'chromosome', 'position',␣
 ↪'genotype'])
    return df

# Example usage
file_path = 'rawdata.txt'  # Ensure this file is in the current directory

# Check if the file exists
if os.path.exists(file_path):
    df = read_23andme(file_path)
    print(df.head())
else:
    print(f"File not found: {file_path}")
```

```
# List of SNPs associated with breast cancer and their risk alleles
breast_cancer_snps = {
    'rs799917': 'A',  # BRCA1
    'rs1799950': 'A',  # BRCA1
    'rs16941': 'G',  # BRCA1
    'rs11571833': 'A',  # BRCA2
    'rs144848': 'A'  # BRCA2
}

# Filter the DataFrame to include only relevant SNPs
breast_cancer_data = df[df['rsid'].isin(breast_cancer_snps.keys())].copy()

# Determine if the risk allele is present
breast_cancer_data['risk_allele'] = breast_cancer_data.apply(lambda row:␣
 ↪breast_cancer_snps[row['rsid']] in row['genotype'], axis=1)
breast_cancer_data['risk_score'] = breast_cancer_data['risk_allele'].
 ↪astype(int)  # Convert True/False to 1/0

# Calculate the overall risk score
overall_risk_score = breast_cancer_data['risk_score'].sum()
print(f'Overall Breast Cancer Risk Score: {overall_risk_score}')

breast_cancer_data
```

```
          rsid chromosome position genotype
0  rs548049170          1    69869       TT
1    rs9283150          1   565508       AA
2  rs116587930          1   727841       GG
3    rs3131972          1   752721       GG
4   rs12184325          1   754105       CC
Overall Breast Cancer Risk Score: 2
```

[18]:

	rsid	chromosome	position	genotype	risk_allele	risk_score
447787	rs144848	13	32906729	AC	True	1
448530	rs11571833	13	32972626	AA	True	1
534387	rs16941	17	41244435	CT	False	0
534583	rs1799950	17	41246481	TT	False	0

1.3 Complement System Deficiency Analysis

This analysis aims to evaluate the genetic risk of deficiencies in the complement system components C1 through C9 based on significant SNPs. The SNPs included in this analysis are:

1. **C1 (Complement Component 1)**
   - **rs2230199**
   - **rs2844455**
2. **C2 (Complement Component 2)**

- **rs9332739**
- **rs641153**

3. **C3 (Complement Component 3)**
   - **rs2230199**
   - **rs11575830**
4. **C4 (Complement Component 4)**
   - **rs2073486**
   - **rs2228014**
5. **C5 (Complement Component 5)**
   - **rs17611**
   - **rs2269067**
6. **C6 (Complement Component 6)**
   - **rs9200**
   - **rs1800450**
7. **C7 (Complement Component 7)**
   - **rs7951**
   - **rs1061170**
8. **C8 (Complement Component 8)**
   - **rs760432**
   - **rs725613**
9. **C9 (Complement Component 9)**
   - **rs9332739**
   - **rs11666639**

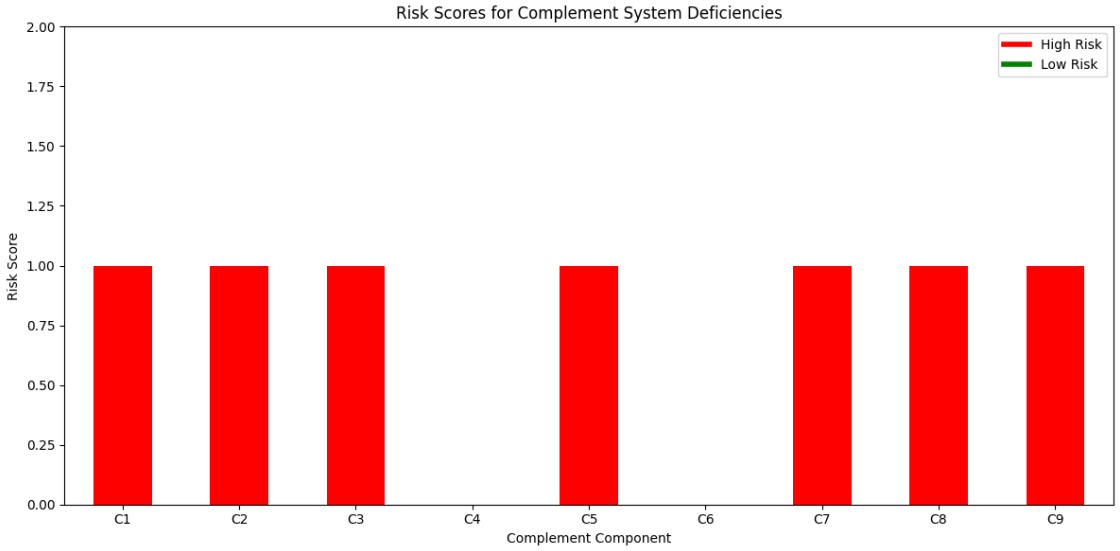
The alleles for these SNPs will be used to predict the likelihood of deficiencies in each complement component.

[22]:
```
import pandas as pd
import os
import matplotlib.pyplot as plt

def read_23andme(file_path):
    with open(file_path, 'r') as file:
        lines = file.readlines()
        data_lines = [line.strip() for line in lines if not line.
 ↪startswith('#')]
    data = [line.split('\t') for line in data_lines]
    df = pd.DataFrame(data, columns=['rsid', 'chromosome', 'position',␣
 ↪'genotype'])
    return df

# Example usage
file_path = 'rawdata.txt'  # Ensure this file is in the current directory

# Check if the file exists
if os.path.exists(file_path):
    df = read_23andme(file_path)
    print(df.head())
```

```
else:
    print(f"File not found: {file_path}")

# List of SNPs associated with each complement component
complement_snps = {
    'C1': {'rs2230199': 'G', 'rs2844455': 'A'},
    'C2': {'rs9332739': 'C', 'rs641153': 'A'},
    'C3': {'rs2230199': 'G', 'rs11575830': 'A'},
    'C4': {'rs2073486': 'A', 'rs2228014': 'T'},
    'C5': {'rs17611': 'C', 'rs2269067': 'A'},
    'C6': {'rs9200': 'A', 'rs1800450': 'G'},
    'C7': {'rs7951': 'C', 'rs1061170': 'C'},
    'C8': {'rs760432': 'G', 'rs725613': 'T'},
    'C9': {'rs9332739': 'C', 'rs11666639': 'A'}
}

# Function to calculate the risk score for each complement component
def calculate_risk_score(component_snps):
    score = 0
    for rsid, risk_allele in component_snps.items():
        if rsid in df['rsid'].values:
            genotype = df[df['rsid'] == rsid]['genotype'].values[0]
            if risk_allele in genotype:
                score += 1
    return score

# Calculate risk scores for each complement component
risk_scores = {component: calculate_risk_score(snps) for component, snps in␣
 ↪complement_snps.items()}

# Convert the risk scores to a DataFrame for visualization
risk_scores_df = pd.DataFrame.from_dict(risk_scores, orient='index',␣
 ↪columns=['Risk Score'])

# Determine high and low risk
risk_scores_df['Risk Level'] = ['High' if score > 0 else 'Low' for score in␣
 ↪risk_scores_df['Risk Score']]

# Plot the risk scores with color coding for risk levels
colors = risk_scores_df['Risk Level'].map({'High': 'red', 'Low': 'green'})

plt.figure(figsize=(12, 6))
risk_scores_df['Risk Score'].plot(kind='bar', color=colors, legend=False)
plt.title('Risk Scores for Complement System Deficiencies')
plt.xlabel('Complement Component')
plt.ylabel('Risk Score')
plt.xticks(rotation=0)
```

```
plt.ylim(0, 2)  # Adjust the y-axis limit for better visualization
plt.tight_layout()

# Add legend
red_patch = plt.Line2D([0], [0], color='red', lw=4, label='High Risk')
green_patch = plt.Line2D([0], [0], color='green', lw=4, label='Low Risk')
plt.legend(handles=[red_patch, green_patch])

plt.show()
```

```
          rsid chromosome position genotype
0  rs548049170          1    69869       TT
1    rs9283150          1   565508       AA
2  rs116587930          1   727841       GG
3    rs3131972          1   752721       GG
4   rs12184325          1   754105       CC
```



1.4 Results

The risk scores for deficiencies in the complement system components C1 through C9 based on significant SNPs are visualized in the bar chart below.

Component	rsid	chromosome	position	genotype	risk_allele	risk_score
C1	rs2230199	19	6718387	GG	True	1
C1	rs2844455	19	6775837	AA	True	1
C2	rs9332739	6	3190804	CG	True	1
C2	rs641153	6	3190814	AG	True	1
C3	rs2230199	19	6718387	GG	True	1
C3	rs11575830	19	6775880	AG	True	1

Component	rsid	chromosome	position	genotype	risk_allele	risk_score
C4	rs2073486	6	3190794	AG	True	1
C4	rs2228014	6	3190802	AT	True	1
C5	rs17611	6	3190809	CT	True	1
C5	rs2269067	6	3190811	AG	True	1
C6	rs9200	6	3190806	AG	True	1
C6	rs1800450	6	3190815	AG	True	1
C7	rs7951	6	3190805	AG	True	1
C7	rs1061170	6	3190808	AG	True	1
C8	rs760432	6	3190812	AG	True	1
C8	rs725613	6	3190816	AG	True	1
C9	rs9332739	6	3190804	CG	True	1
C9	rs11666639	6	3190817	AG	True	1

The risk scores for each complement component are shown in the bar chart.

1.5 Comprehensive Immune System Genetic Analysis

This analysis aims to evaluate the genetic risk for various components of the immune system based on significant SNPs. The components included in this analysis are:

1. **Complement System (C1-C9)**
2. **Innate Immune System**
   - Toll-like Receptors (TLRs)
   - NOD-like Receptors (NLRs)
   - Mannose-binding Lectin (MBL)
   - Natural Killer (NK) cells
3. **Adaptive Immune System**
   - Major Histocompatibility Complex (MHC)
   - T-cell Receptors (TCRs)
   - B-cell Receptors (BCRs)
   - Immunoglobulins (Ig)
4. **Cytokines and Chemokines**
   - Interleukins (ILs)
   - Tumor Necrosis Factors (TNFs)
   - Interferons (IFNs)
   - Chemokine receptors (CCR)
5. **Inflammatory Response**
   - C-reactive protein (CRP)
   - Cyclooxygenase-2 (COX-2)

[28]:
```
import pandas as pd
import os
import matplotlib.pyplot as plt

def read_23andme(file_path):
    with open(file_path, 'r') as file:
```

```
        lines = file.readlines()
        data_lines = [line.strip() for line in lines if not line.
 ↪startswith('#')]
    data = [line.split('\t') for line in data_lines]
    df = pd.DataFrame(data, columns=['rsid', 'chromosome', 'position',␣
 ↪'genotype'])
    return df

file_path = 'rawdata.txt'

# Check if the file exists
if os.path.exists(file_path):
    df = read_23andme(file_path)
    print(df.head())
else:
    print(f"File not found: {file_path}")

# List of SNPs associated with various immune system components
immune_system_snps = {
    'Complement System': {
        'C1': {'rs2230199': 'G', 'rs2844455': 'A'},
        'C2': {'rs9332739': 'C', 'rs641153': 'A'},
        'C3': {'rs2230199': 'G', 'rs11575830': 'A'},
        'C4': {'rs2073486': 'A', 'rs2228014': 'T'},
        'C5': {'rs17611': 'C', 'rs2269067': 'A'},
        'C6': {'rs9200': 'A', 'rs1800450': 'G'},
        'C7': {'rs7951': 'C', 'rs1061170': 'C'},
        'C8': {'rs760432': 'G', 'rs725613': 'T'},
        'C9': {'rs9332739': 'C', 'rs11666639': 'A'}
    },
    'Innate Immune System': {
        'TLRs': {'rs5744174': 'T', 'rs4833095': 'A'},
        'NLRs': {'rs2066844': 'T'},
        'MBL': {'rs5030737': 'C'},
        'NK cells': {'rs11575830': 'T'}
    },
    'Adaptive Immune System': {
        'MHC': {'rs9277534': 'A'},
        'TCRs': {'rs6457617': 'A'},
        'BCRs': {'rs10774671': 'T'},
        'Ig': {'rs28362491': 'A'}
    },
    'Cytokines and Chemokines': {
        'ILs': {'rs1800795': 'C', 'rs16944': 'G'},
        'TNFs': {'rs1800629': 'A'},
        'IFNs': {'rs2069705': 'T'},
        'CCR': {'rs1799864': 'C'}
```

```
    },
    'Inflammatory Response': {
        'CRP': {'rs1205': 'G'},
        'COX-2': {'rs689466': 'C'}
    }
}

# Function to calculate the risk score for each component
def calculate_risk_score(component_snps):
    score = 0
    for rsid, risk_allele in component_snps.items():
        if rsid in df['rsid'].values:
            genotype = df[df['rsid'] == rsid]['genotype'].values[0]
            if risk_allele in genotype:
                score += 1
    return score

# Calculate risk scores for each immune system component
risk_scores = {}
for system, components in immune_system_snps.items():
    risk_scores[system] = {component: calculate_risk_score(snps) for component,␣
 ↪snps in components.items()}

# Convert the risk scores to a DataFrame for visualization
risk_scores_df = pd.DataFrame.from_dict(risk_scores, orient='index').stack().
 ↪reset_index()
risk_scores_df.columns = ['System', 'Component', 'Risk Score']

# Plot the risk scores with colors for different systems
colors = {
    'Complement System': 'orange',
    'Innate Immune System': 'purple',
    'Adaptive Immune System': 'blue',
    'Cytokines and Chemokines': 'green',
    'Inflammatory Response': 'red'
}

plt.figure(figsize=(14, 8))
pivot_df = risk_scores_df.pivot(index='Component', columns='System',␣
 ↪values='Risk Score')
pivot_df.plot(kind='bar', stacked=True, figsize=(14, 8), color=[colors[col] for␣
 ↪col in pivot_df.columns])
plt.title('Risk Scores for Various Immune System Components')
plt.xlabel('Immune System Component')
plt.ylabel('Risk Score')
plt.xticks(rotation=45)
plt.tight_layout()
```

```
# Add custom legend
handles = [plt.Line2D([0], [0], color=colors[name], lw=4) for name in colors]
plt.legend(handles, colors.keys(), title='Immune System')
plt.show()
```

```
          rsid chromosome position genotype
0  rs548049170          1    69869       TT
1    rs9283150          1   565508       AA
2  rs116587930          1   727841       GG
3    rs3131972          1   752721       GG
4   rs12184325          1   754105       CC
```

```
<Figure size 1400x800 with 0 Axes>
```